

Hemoglobinopathy Fact Sheet

Hemoglobin C

Hemoglobin C* is an inherited variant of normal adult hemoglobin (hemoglobin A). It results from a single change in one of the hundreds of *amino acids* that make up hemoglobin. Hemoglobin C is relatively common, particularly among people of African heritage. In the United States about 1 of 50 (2%) of African Americans has a hemoglobin C gene. Other groups where it is found include Italians (especially Sicilians), Greeks, Syrians, Turks, Cypriots, and Asiatic Indians. Summarized below are the three most commonly encountered hemoglobin patterns that involve hemoglobin C. *Genetic counseling* is advisable for families affected by these conditions to promote understanding of the significance for them or for future offspring.

Hemoglobin C Trait (*phenotype* FAC in infants and AC in adults)

Hemoglobin C trait results when a person inherits a hemoglobin C *gene* from one parent and a hemoglobin A gene from the other parent. This does not cause any health problems. For an infant identified with hemoglobin C trait on two newborn screening specimens, no further testing is indicated. However, other family members may be interested in having their individual phenotypes determined to learn if they have hemoglobin patterns involving hemoglobin C that can cause health problems for themselves or future children.

Hemoglobin C Disease (*phenotype* FC in infants and CC in adults)

Hemoglobin C disease results when a person inherits only hemoglobin C *genes*, one from each parent. A person with hemoglobin C disease has no genes for hemoglobin A. This condition causes few health problems, however, mild to moderate *hemolytic anemia* may develop. For an infant identified with hemoglobin C disease by newborn screening, the findings should be confirmed by analysis of blood *indices* and determination of hemoglobin phenotype.

Hemoglobin SC Disease (*phenotype* FSC in infants and SC in adults)

Sickle C disease is a type of sickle cell disease. (In the United States about 1 of 1,000 African Americans has sickle C disease.) It occurs when a person inherits a *gene* for sickle hemoglobin (hemoglobin S) from one parent and a gene for hemoglobin C from the other parent. The red blood cells will then sometimes change from a very flexible round shape into a rigid crescent or “sickle” shape. Sickle shaped red blood cells can prevent the usual flow of blood and oxygen to body organs. Those affected with sickle C disease can have any of the health problems that individuals with sickle cell anemia (two genes for hemoglobin S) can have, though these problems tend to be less severe and occur less frequently. Sickle C disease, however, can have its own significant complications, such as retinal detachment, painful episodes, and aseptic necrosis of the hips. These children should be treated according to the sickle cell disease protocol, although these children may not have an increased risk of overwhelming infection.

** This and all other italicized words are defined on the reverse side of this sheet.*



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DEFINITIONS of *italicized* words

Hemoglobin - the proteins in red blood cells that carries oxygen to body tissues.

Amino acids - building blocks of proteins.

Genetic counseling - meeting with someone knowledgeable in genetics to discuss information and risks about an inherited condition.

Phenotype - a characteristic produced by a set of genes (a gene set is called a genotype).

Gene - a unit of inheritance that codes for a specific protein.

Hemolytic anemia - a type of anemia in which red blood cells are more fragile than usual and have a shorter life span.

Indices - set of values used to describe red blood cell characteristics.